



The MyNeuro genetic test analyses a set of markers related to biological processes that influence brain function and mood. processes that influence brain function and mood. The cell signalling process, the cell signalling, the individual's response to stress, Wnt signalling, inflammation and methylation, in order to understand how genetic variations in these processes can compromise neuronal homeostasis and predispose to certain neuropsychiatric disorders.

Genes related to neurotransmitters and their receptors, such as dopamine, serotonin and GABA, have also been analysed. Neurotransmitters play a fundamental role in communication between nerve cells, mood regulation and cognitive processes. This report aims to provide more accurate and personalised information on the patient's predisposition to mood alterations and addictive behaviours.

These biological processes have been grouped into four different categories:





# **SUMMARY OF RESULTS:**

1. Emotional homeostasis



2. Behavioural regulation



3. Addictive behaviour



4. Neuroinflammation





## RECOMMENDATIONS

**Dopamine:** Diet rich in magnesium, tyrosine, B vitamins, especially B5 and B6, vitamin D and omega-3. Recommended foods include bananas, oranges, apples, tomato, spinach, aubergine, beans, peas, chicken and almonds.

**Serotonine:** Physical activity, stress management techniques and consumption of tryptophan, which is found in foods such as cheese, chicken, egg whites, fish and milk.

**GABAERGIC:** Mayor consumo de piridoxina (vitamina B6), presente en alimentos como crucíferas, legumbres, tomate, espinacas, champiñones y cereales integrales.

**Endocannabinoids:** Increased intake of pyridoxine (vitamin B6), found in foods such as cruciferous vegetables, legumes, tomatoes, spinach, mushrooms and whole grains.

**Cell signalling:** Consumption of foods rich in folic acid, omega-3. foods rich in antioxidants, calcium and vitamins.

**Stress response:** Follow techniques that help to better control stress. Consumption of foods such as oily fish, dark chocolate, turmeric, olive oil, bananas, garlic or flax seeds as they reduce cortisol and help to combat stress.

**Methylation:** Adequate intake of folic acid (vitamin B9), especially found in green leafy vegetables and legumes.

**Neuroinflammation:** A diet with a balanced omega-3/omega-6 ratio, increasing the consumption of foods rich in omega-3, such as oily fish and seafood, nuts and vegetable oils. Seasoning meals with spices such as ginger, cinnamon, turmeric or garlic for their anti-inflammatory properties and consuming polyphenols, which are present in fruit, vegetables, tea and cocoa.



# YOUR RESULTS IN DETAIL

# 1. EMOTIONAL HOMEOSTASIS



### 1.1. Information about emotional homeostasis

Processes that compromise the dynamic balance and stability of the body's emotional responses to ensure healthy emotional functioning.

## 1.2. Impact of associated processes

Process		Risk
DOPAMINE	LOW	
SEROTONINE	HIGH	
ENDOCANNABINOID	LOW	
CELL SIGNALLING	HIGH	
SIGNALLING WNT	NEUTRAL	
STRESS RESPONSE	LOW	
METILATION	LOW	

Biological process	Gene	Variante	Reference genotype	Patient genotype	Risk
Dopaminergic	DRD1	rs4532	TT	СТ	
Dopaminergic	DRD1	rs5326	СС	CC	
Dopaminergic	DRD2	rs1800497	CC	GG	
Dopaminergic	DRD3	rs6280	CC	TT	
Dopaminergic	DRD4	rs1800955	TT	TC	
Dopaminergic	COMT	rs4680	GG	AA	
Dopaminergic	OPRM1	rs1799971	AA	AA	
Serotoninergic	1A-HTR1A	rs6295	CC	CG	



Serotoninergic	SLC6A4	rs1042173	AA	CC	
Endocannabinoids	CNR1	rs2023239	TT	TT	
Endocannabinoids	FAAH	rs324420	CC	CC	
Cell signalling	AKT1	rs2494732	TT	TC	
Cell signalling	ANK3	rs1938526	AA	AA	
Cell signalling	ANK4	rs10994336	CC	CC	
Cell signalling	BDNF	rs6265	CC	CC	
Cell signalling	CACNAI	rs1006737	GG	AA	
Cell signalling	CHRNA3	rs16969968	GG	GA	



# 2. BEHAVIOURAL REGULATION



# 2.1. Information about behavioural regulation

A set of biological processes that influence an individual's actions and behavioural responses.

## 2.2. Impact of associated processes

Process		Risk
DOPAMINE	LOW	
SEROTONINE	HIGH	
CELL SIGNALLING	HIGH	

Biological process	Gene	Variante	Reference genotype	Patient genotype	Risk
Dopaminergic	DRD1	rs4532	TT	СТ	
Dopaminergic	DRD1	rs5326	CC	CC	
Dopaminergic	DRD2	rs1800497	CC	GG	
Dopaminergic	DRD3	rs6280	CC	TT	
Dopaminergic	DRD4	rs1800955	TT	TC	
Dopaminergic	COMT	rs4680	GG	AA	
Dopaminergic	OPRM1	rs1799971	AA	AA	
Serotoninergic	1A-HTR1A	rs6295	CC	CG	
Serotoninergic	SLC6A4	rs1042173	AA	CC	
Cell signalling	AKT1	rs2494732	TT	TC	
Cell signalling	ANK3	rs1938526	AA	AA	
Cell signalling	ANK4	rs10994336	CC	CC	
Cell signalling	BDNF	rs6265	CC	CC	
Cell signalling	CACNAI	rs1006737	GG	AA	
Cell signalling	CHRNA3	rs16969968	GG	GA	



# 3. ADDICTIVE BEHAVIOUR



### 3.1. Information about addictive behaviours

Repetitive and compulsive behaviour patterns that are linked to substance seeking and use.

# 3.2. Impact of associated processes

Process		Risk
DOPAMINE	LOW	
SEROTONINE	HIGH	
ENDOCANNABINOID	LOW	
GABA	HIGH	
CELL SIGNALLING	HIGH	

Biological process	Gene	Variante	Reference genotype	Patient genotype	Risk
Dopaminergic	DRD1	rs4532	TT	СТ	
Dopaminergic	DRD1	rs5326	CC	CC	
Dopaminergic	DRD2	rs1800497	CC	GG	
Dopaminergic	DRD3	rs6280	CC	TT	
Dopaminergic	DRD4	rs1800955	TT	TC	
Dopaminergic	COMT	rs4680	GG	AA	
Dopaminergic	OPRM1	rs1799971	AA	AA	
Serotoninergic	1A-HTR1A	rs6295	CC	CG	
Serotoninergic	SLC6A4	rs1042173	AA	CC	
Endocannabinoids	CNR1	rs2023239	TT	TT	
Endocannabinoids	FAAH	rs324420	CC	CC	
GABAérgico	GABRA2	rs279858	TT	TC	
Cell signalling	AKT1	rs2494732	TT	TC	
Cell signalling	ANK3	rs1938526	AA	AA	
Cell signalling	ANK4	rs10994336	CC	CC	
Cell signalling	BDNF	rs6265	CC	CC	
Cell signalling	CACNAI	rs1006737	GG	AA	
Cell signalling	CHRNA3	rs16969968	GG	GA	



# 4. **NEUROINFLAMMATION**



## 4.1. Information about neuroinflammation

Conditions of the nervous system characterised by a progressive loss of neural structures and functions.

# 4.2. Impact of associated processes

Process		Risk
NEUROINFLAMMATION	NEUTRAL	

Biological process	Gene	Variante	Reference genotype	Patient genotype	Risk
Neuroinflammation	IL1A	rs17561	GG	CA	
Neuroinflammation	IL1A	rs1800587	CC	GA	
Neuroinflammation	IL1B	rs1143634	CC	GG	
Neuroinflammation	IL1B	rs16944	AA	GG	
Neuroinflammation	ILIRN	rs419598	CC	TT	
Neuroinflammation	IL-6	rs1800795	GG	CG	
Neuroinflammation	IL-10	rs1800896	GG	TC	
Neuroinflammation	TNFA	rs1800629	GG	GG	
Neuroinflammation	CD33	rs3865444	TT	CC	000
Neuroinflammation	MS4A	rs744373	TT	AA	
Neuroinflammation	TREM2	rs143332484	CC	CC	
Neuroinflammation	CRP	rs1205	CC	TT	

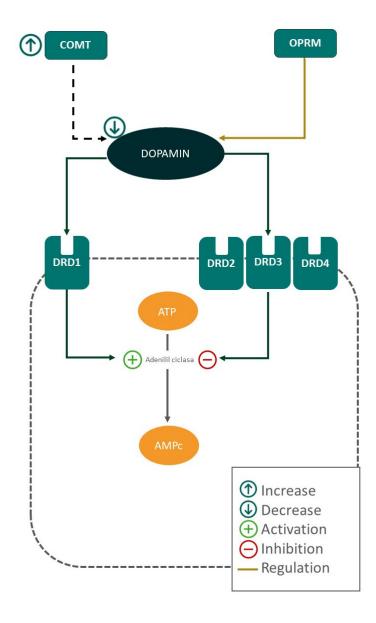


### **ANNEX I: DETAILS OF PHYSIOLOGICAL PROCESSES**

### 1. DOPAMINE

#### **GLOBAL RISK: LOW**

Dopamine is a neurotransmitter, a chemical found in the brain and central nervous system that plays a key role in communication between nerve cells or neurons. Dopamine plays several roles in the brain and body, being involved in a wide variety of processes, including movement control, mood regulation, motivation, pleasure and decision-making, so alterations in its production can affect mood.





### DRD1 rs4532 | rs5326

This gene encodes the dopamine D1 receptor, a G protein-coupled receptor that stimulates adenylyl cyclase, an intracellular enzyme that catalyses the conversion of ATP to cAMP, acting as a second messenger. D1 receptors regulate neuronal growth and development and mediate some behavioural responses. The T allele inhibits translation of the receptor, thus affecting its functionality. The patient is at low risk (rs4532). and has no rs5326-related risk for increased susceptibility to alcohol consumption, smoking and impulsive behaviour.

Patient genotype: **CT | CC** 

Ri	sk:

#### DRD2 rs1800497

Este gen codifica el receptor D2 de la dopamina. Este receptor acoplado a proteína G inhibe la adenilil ciclasa. The CC genotype is associated with normal DRD2 activity and therefore does not pose an increased risk to the patient.

Patient genotype: **GG** 

	Ri	sk:	
$\equiv$	$\supset$ $\subset$	$\supset$ $\subset$	

#### **DRD3 rs6280**

This gene encodes the dopamine D3 receptor. This G protein-coupled receptor inhibits adenylyl cyclase. It is a receptor located in the limbic area of the brain, which is related to cognitive, emotional and endocrine functions. The TT genotype is associated with normal DRD3 activity and therefore does not pose an increased risk to the patient.

Patient genotype:

TT

R	isk:

#### DRD4 rs1800955

This gene encodes the D4 dopamine receptor. This G protein-coupled receptor inhibits adenylyl cyclase. It is a receptor located in the limbic area of the brain, which is related to cognitive, emotional and endocrine functions. which is related to cognitive, emotional and endocrine functions. The C allele in this gene is associated with higher transcript levels of DRD4, thus a low risk for the development of attention deficit disorder and a greater predisposition to novelty seeking.

Patient genotype:

TC



#### **COMT rs4680**

Catechol-O-methyltransferase is an enzyme encoded by the COMT gene that catalyses the transfer of a methyl group from S-adenosylmethionine to the catecholamines the transfer of a methyl group from S-adenosylmethionine to catecholamines, including the neurotransmitters dopamine, epinephrine and norepinephrine, leading to one of the main degradation pathways for catecholamine transmitters. The A allele is associated with reduced COMT activity and thus higher dopamine levels and the patient is at medium risk of developing reduced tolerance to pain and stress.

Patient genotype:

Risk:

#### **OPRM1 rs1799971**

This gene encodes one of the three opioid receptors in the body. MOR is the main target for endogenous opioid peptides and opioid analgesic agents such as beta-endorphin and enkephalins. It plays an important role in the dependence on other drugs of abuse, such as nicotine, cocaine and alcohol, through its modulation of the dopaminergic system. The AA genotype is associated with normal OPRM1 activity and therefore does not pose an increased risk to the patient.

Patient genotype:

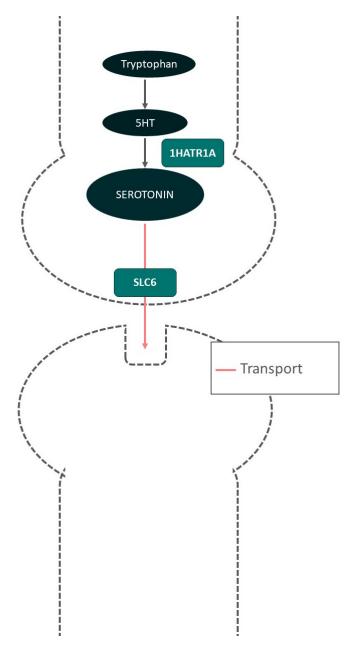
AA



#### 2. SEROTONIN

#### **GLOBAL RISK:** HIGH

Serotonin is a neurotransmitter that generally acts as an inhibitor in the central nervous system. nervous system. The precursor for its synthesis is tryptophan, which is transported through the blood to the brain where it is uptaken by nerve blood to the brain where it is taken up by nerve terminals and converted to 5-hydroxytryptophan. hydroxytryptophan (5-HTP) by the enzyme tryptophan hydroxylase. Serotonin is involved in learning, memory, happiness and sleep. Low levels of levels have been linked to depression, anxiety, sleep problems and even gastrointestinal conditions. gastrointestinal conditions.





#### 1A-HTR1A rs6295

This gene encodes a G protein-coupled receptor for 5-hydroxytryptamine. The G allele has been associated with increased 5-HT(1A) receptor concentration in presynaptic neurons and reduced neuronal firing, thus posing a medium risk for the development of depression.

Patient genotype:

CG

Risk:



#### SLC6A4 rs1042173

This gene encodes a membrane protein that transports serotonin from synaptic spaces to presynaptic neurons. The encoded protein terminates the action of serotonin and recycles it in a sodium-dependent manner. This protein is a target of psychomotor stimulants, such as amphetamines and cocaine. The C allele is associated with increased expression of SLC6A4, and thus poses a medium risk for increased susceptibility to alcohol consumption.

Patient genotype:

CC

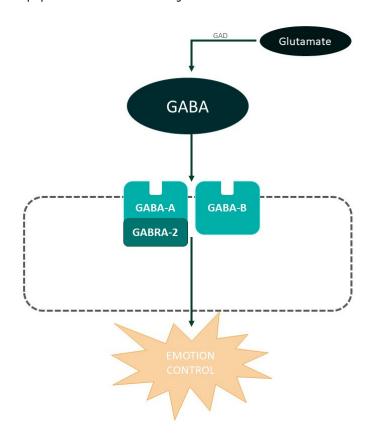




#### 3. GABA

#### **GLOBAL RISK: HIGH**

Gamma-aminobutyric acid (GABA) is an inhibitory neurotransmitter of the central nervous system. It is synthesised from glutamic acid, another excitatory neurotransmitter in the brain. The enzyme glutamate decarboxylase (GAD) converts glutamate to GABA, which will be detected by GABA-A and GABA-B receptors on nerve cells, thereby reducing the excitability of the cell. This pathway is involved in the regulation of muscle tone, inhibition of neuronal excitability and regulation of mood and anxiety, so that stimulation of the GABAergic system causes a state of sedation, amnesia or even ataxia, while inhibition leads to sleep problems or anxiety.





#### **GABRA2 rs279858**

The protein encoded by this gene is a member of the immunophilin family of proteins, which play a role in immunoregulation and basic cellular processes involving protein folding and trafficking. It interacts functionally with mature hetero-oligomeric progesterone receptor complexes. The C allele has been associated with a poorer response of the body to alcohol, thus a medium risk to respond more slowly to the effects of alcohol and thus a higher predisposition of the patient to alcoholism.

Patient genotype:

TC

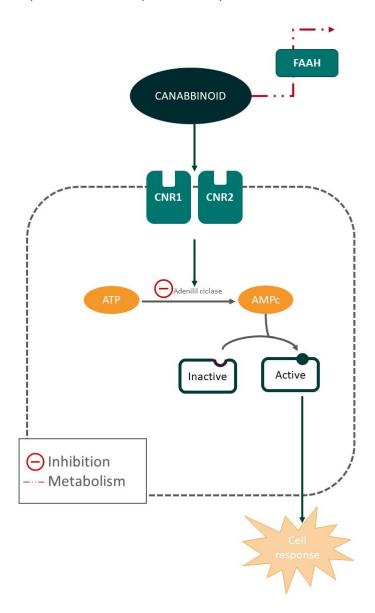


### 4. ENDOCANNABINOIDS

#### **GLOBAL RISK: LOW**

The main receptors within the Endocannabinoid system are the CB1 and CB2 receptors, transmembrane proteins capable of transmitting extracellular signals into the cell when cannabinoids bind to them, inhibiting the release of neurotransmitters such as GABA or glutamate.

The endocannabionoid system is involved in learning and memory processes, emotions, addictions, as well as pain and neuroprotective processes.





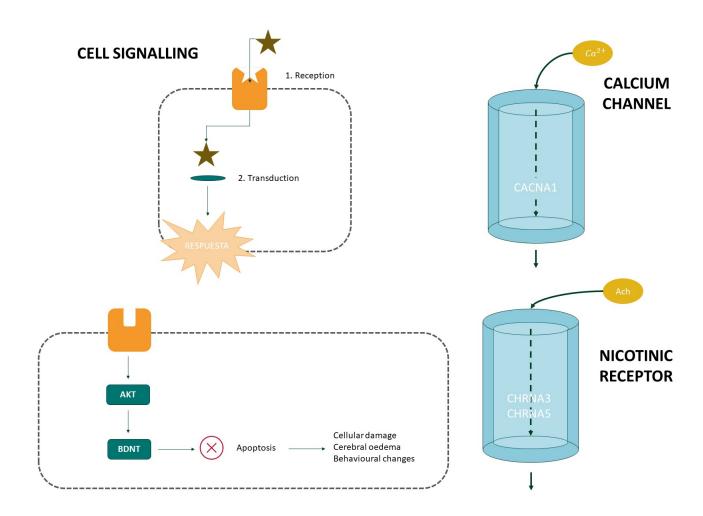
## CNR1 rs2023239 This gene encodes one of two cannabinoid receptors. These recep-Patient genotype: tors are members of the G-protein-coupled receptor family, which inhibit adenylate cyclase activity in a dose-dependent, stereoselective and pertussis toxin-sensitive manner. They are implicated in cannabinoid-induced central nervous system effects such as alterations in mood and cognition. The TT genotype has been associated Risk: with normal CNR1 function and therefore does not pose an increased risk to the patient. **FAAH rs324420** Patient genotype: This gene encodes a protein responsible for the hydrolysis of several CC primary and secondary fatty acid amides, including the neuromodulatory compounds anandamide and oleamide. The CC genotype has been associated with normal FAAH activity and therefore does not pose an increased risk to the patient. Risk:



### 5. CELL SIGNALLING

#### **GLOBAL RISK: HIGH**

Cell signalling is the process by which the cell responds to substances such as hormones or neurotransmitters when they bind to their receptors. Signals are transmitted from one cell to another producing a specific cellular response. Normal cell signalling is important for proper communication between neurons and neuronal survival. Disturbances in these pathways affect memory and lead to mood disturbances, for example in bipolar disorder.





## FAAH rs324420 Patient genotype: This gene encodes a protein responsible for the hydrolysis of several CC primary and secondary fatty acid amides, including the neuromodulatory compounds anandamide and oleamide. The CC genotype has been associated with normal FAAH activity and therefore does not pose an increased risk to the patient. Risk: AKT1 rs2494732 This gene encodes one of three members of the AKT family Patient genotype: of serine/threonine protein kinases, which are phosphorylated by TC phosphoinositide 3-kinase. AKT/PI3K is a key component of many signalling pathways involving membrane ligand binding, regulating cellular functions such as cell proliferation, survival, metabolism and angiogenesis. The C allele is associated with an altered function of the AKT1 gene and therefore poses a low risk for the development of psychotic behavior. ANK3 rs1938526 Patient genotype: The ankyrins are a family of proteins that play key roles in activities AA such as cell motility, activation, proliferation, contact and maintenance of specialised membrane domains. The AA genotype has been associated with normal cell signaling, so it does not pose an increased risk to the patient. Risk: ANK4 rs10994336 Patient genotype: The ankyrins are a family of proteins that play key roles in activities CC such as cell motility, activation, proliferation, contact and maintenance of specialised membrane domains. The CC genotype has been associated with normal data in the axiom, so it does not pose an increased risk to the patient. Risk:



#### **BDNF** rs6265

This gene encodes a member of the nerve growth factor family of proteins. Binding of this protein to its associated receptor promotes neuronal survival in the adult brain. This gene may play a role in the regulation of the stress response and in the biology of mood disorders. The CC genotype has been associated with normal BDNF function, so it does not pose an increased risk to the patient.

Patient genotype:

CC

Risk:

#### **CACNA1 rs1006737**

This gene encodes an alpha-1 subunit of a voltage-dependent calcium channel. Calcium channels mediate the entry of calcium ions into the cell following membrane polarisation. The A allele is associated with an alteration of the gene and therefore poses a medium risk for the development of depression, schizophrenia and bipolar disorder.

Patient genotype:

AA

Risk:



### CHRNA3 | CHRNA5 rs16969968

This locus encodes a member of the nicotinic acetylcholine receptor family of proteins. This locus encodes an alpha-type subunit, as it contains characteristic adjacent cysteine residues. The encoded protein is a ligand-activated ion channel that plays a role in neurotransmission. The A allele is associated with enhanced pleasurable responses and therefore the patient is at low risk of developing dependence on addictive substances such as nicotine.

Patient genotype:

GA

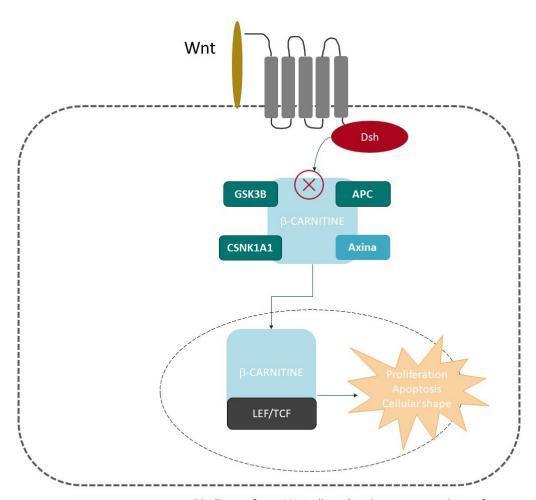




### 6. Wnt SIGNALLING

#### **GLOBAL RISK: NEUTRAL**

Wnt signalling is a highly conserved cell signalling pathway essential for both the homeostasis of the organism and the regulation of processes such as cell proliferation and differentiation. It involves the accumulation of the protein beta-catenin in the cell nucleus, acting as a transcription factor for the regulation of gene expression involved in cell proliferation and development.



Binding of a Wnt ligand triggers a series of intracellular signals that lead to destabilisation of the multi-protein destruction complex. This allows cytosolic accumulation of  $\beta$ -catenin and its migration to the nucleus, where it binds to LEF/TCF factors. Transcription of the pathway target genes from the LEF/TCF/ $\beta$ -catenin complex then occurs.



### APC rs2546110 | rs3846716

This gene encodes a tumour suppressor protein that acts as an antagonist of the Wnt signalling pathway, as well as being involved in other processes such as cell migration and adhesion, transcriptional activation and apoptosis. The G allele is associated with altered APC gene function. The patient has a medium risk associated with rs2546110. and a medium risk for rs3846716 in the development of psychotic behavior.

Patient genotype:

GG | GG



### GSK3B rs334555 | rs11925868

The protein encoded by this gene is a member of the immunophilin family of proteins, which play a role in immunoregulation and basic cellular processes involving protein folding and trafficking. The C allele has been associated with an alteration in the regulation of GSK3B, and therefore poses a medium risk for the rs334555 marker. and a low risk for rs11925868 in the development of mood disorders such as depression or bipolar disorder.

Patient genotype: **CC | CA** 



#### CSNK1A1 rs10045427

Enables protein serine/threonine kinase activity. Involved in several processes, including negative regulation of the canonical Wnt signalling pathway, peptidyl-serine phosphorylation and positive regulation of ubiquitin-dependent protein proteasomal catabolic processing. The C allele is associated with an alteration in CSNK1A1 activity and therefore poses a low risk for the development of mood disorders for the patient.

Patient genotype: **AC** 

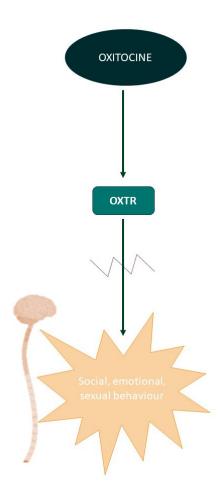




### 7. STRESS RESPONSE

#### **GLOBAL RISK: LOW**

Stress response is a set of physiological and behavioural responses that the body experiences when faced with stressful external stimuli. It is a response necessary for adaptation to the environment. It is mediated by different systems such as the autonomic nervous system, the endocrine system, the immune system or the central nervous system. When the body faces chronic stress, it can lead to mood disorders such as anxiety or depression.





#### FKBP5 rs1360780

The protein encoded by this gene is a member of the immunophilin family of proteins, which play a role in immunoregulation and basic cellular processes involving protein folding and trafficking. This encoded protein is a cis-trans prolyl isomerase that binds to the immunosuppressants FK506 and rapamycin. It is thought to mediate calcineurin inhibition. The FKBP5 genotype has been associated with normal FKBP5 activity and therefore does not pose an increased risk to the patient.

Patient genotype: **CC** 

Risk:

#### **OXTR** rs53576

The protein encoded by this gene belongs to the G protein-coupled receptor family and acts as a receptor for oxytocin. Its activity is mediated by G proteins that activate a phosphatidylinositol-calcium second messenger system. The FKBP5 genotype has been associated with normal FKBP5 activity and therefore does not pose an increased risk to the patient.

Patient genotype:

AG

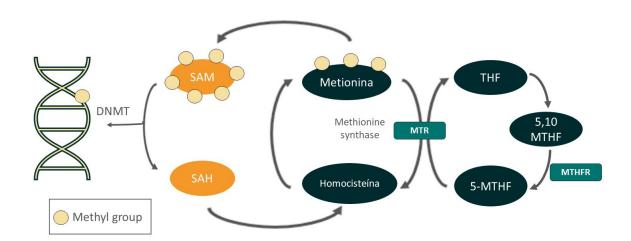




### 8. METHYLATION

#### **GLOBAL RISK: LOW**

Methylation is a process in which methyl groups are added to molecules such as DNA, proteins or lipids. It is of great relevance in the nervous system, as it is involved in the synthesis and regulation of neurotransmitters. Alterations in methylation processes can lead to dysregulation in the synthesis or uptake of neurotransmitters, which can contribute to mood disorders such as depression and anxiety.





### MTHFR rs1801131 | rs1801133

The protein encoded by this gene catalyses the conversion of 5,10-methylenetetrahydrofolate to 5-methyltetrahydrofolate, a cosubstrate for the remethylation of homocysteine to methionine. The T (rs1801131) and G (rs1801133) alleles have been associated with reduced MTHFR activity, thus a high risk for the rs1801131 marker y y has no rs1801133-related risk for increased mood disorders in the patient.

Patient genotype: **TT | AA** 

Risk:

#### MTR rs1805087

This gene encodes 5-methyltetrahydrofolate-homocysteine methyltransferase. This enzyme, also known as cobalamin-dependent methionine synthase, catalyses the final step in methionine biosynthesis. The AA genotype has been associated with normal enzyme capacity and therefore does not pose an increased risk to the patient.

Patient genotype:



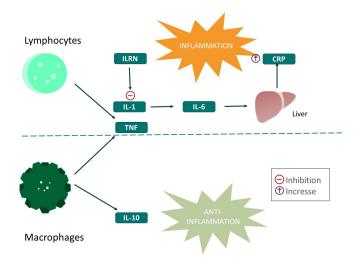
#### 9. NEUROINFLAMMATION

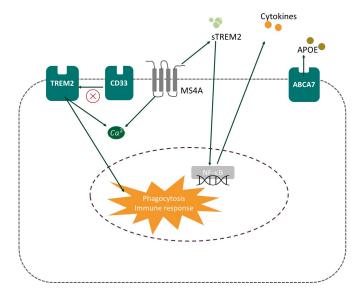
#### **GLOBAL RISK: NEUTRAL**

Neuroinflammation is a biological process involving an inflammatory response in the central nervous system, which comprises the brain and spinal cord. This process can be triggered by a variety of causes, such as infection, injury, autoimmune disorders or oxidative stress.

It is a process mediated primarily by microglia and astrocytes, which release cytokines and other inflammatory molecules that may contribute to neuronal degeneration and impaired brain function.

Un estado de inflamación prolongado en el sistema nervioso central puede alterar los sistemas de señalización y alterar los mecanismos que regulan el estado de ánimo, pudiendo ser el causante de trastornos psiquiátricos como la depresión y ansiedad, además de estar relacionado con enfermedades neurodegenerativas como el Alzheimer o Parkinson.







### IL1A rs17561 | rs1800587

The protein encoded by this gene is a member of the interleukin 1 family of cytokines, a pleiotropic cytokine involved in various immune responses, inflammatory processes and haematopoiesis. The A allele has been associated with increased IL1-A activity, and therefore poses a medium risk for the rs17561 marker. and a medium risk for rs1800567 in the development of neuroinflammatory disorders, mood disorders and depression in the patient.

Patient genotype: **CA | GA** 



## IL1B rs1143634 | rs16944

The protein encoded by this gene is a member of the interleukin I family of cytokines, a pleiotropic cytokine involved in various immune responses, inflammatory processes and haematopoiesis. The A (rs1143634) and G (rs16944) alleles have been associated with increased ILI-B activity and therefore pose a high risk for the rs1143634 marker and a high risk for rs16944 for the development of neuroinflammatory disorders, mood disorders and depression in the patient.

Patient genotype:

GG | GG



#### **IL1RN rs419598**

The protein encoded by this gene is a member of the interleukin-1 cytokine family. This cytokine is an important mediator of the inflammatory response and is involved in various cellular activities such as cell proliferation, differentiation and apoptosis. The T allele has been associated with increased IL1-RN activity and therefore poses a medium risk for the development of neuroinflammatory disorders, mood disorders and depression in the patient.

Patient genotype:

TT



#### IL-6 rs1800795

This gene encodes a cytokine involved in inflammation and B-lymphocyte maturation. The protein is mainly produced at sites of acute and chronic inflammation, where it is secreted into serum and induces a transcriptional inflammatory response via the interleukin-6 receptor. The C allele has been associated with lower levels of IL-6 and therefore the patient is at low risk of developing Alzheimer's disease.

Patient genotype:

CG





#### IL-10 rs1800896

This gene encodes a cytokine involved in inflammation and the maturation of B-lymphocytes, which has an anti-inflammatory effect on the body. The T allele has been associated with reduced IL10 levels and therefore poses a low risk for the development of neuroinflammatory disorders, mood disorders and depression in the patient.

Patient genotype:

TC

Risk:



#### TNFA rs1800629

This gene encodes a multifunctional proinflammatory cytokine that belongs to the tumour necrosis factor (TNF) superfamily. This cytokine is mainly secreted by macrophages. It can bind to its receptors TNFRSF1A/TNFR1 and TNFRSF1B/TNFBR and thus function through them. The GG genotype has been associated with normal TNFA expression and therefore does not pose an increased risk to the patient.

Patient genotype:

GG

Risk:



#### CD33 rs3865444

This gene activates binding to protein phosphatase and sialic acid. It is involved in processes such as negative regulation of cytokine production and monocyte activation, as well as positive regulation of protein tyrosine phosphatase activity. The CC genotype has been associated with normal CD33 activity and therefore does not pose an increased risk to the patient.

Patient genotype:

CC

Risk:



#### MS4A rs744373

It encodes a domain of membrane proteins that express unique expression patterns between hematopoietic cells and non-lymphoid tissues. It also encodes a surface molecule of B lymphocytes that plays a role in the development and differentiation into plasma cells. The AA genotype has been associated with normal MS4A activity and therefore does not pose an increased risk to the patient.

Patient genotype:

AA





TREM2 rs143332484		
This gene encodes a membrane protein that forms a signaling receptor complex with tyrosine kinase binding protein. The encoded protein acts during an immune response and may be involved in	Patient genotype: <b>CC</b>	
chronic inflammation by triggering cytokine production. The CC genotype has been associated with normal TREM2 expression and is therefore not an increased risk for the patient.	Risk:	
CRP rs1205		
CRP rs1205  The protein encoded by this gene belongs to the pentraxin family, involved in complement activation and amplification through communication with initiation pattern recognition molecules, but also in	Patient genotype: <b>TT</b>	



# **SUMMARY OF RESULTS**

The following table shows the variants detected in the patient and their impact:

Biological process	Gene	Variant	Reference genotype	Patient genotype	Risk
Dopaminergic	DRD1	rs4532	TT	СТ	
Dopaminergic	DRD1	rs5326	CC	CC	
Dopaminergic	DRD2	rs1800497	CC	GG	
Dopaminergic	DRD3	rs6280	CC	TT	
Dopaminergic	DRD4	rs1800955	TT	TC	
Dopaminergic	COMT	rs4680	GG	AA	
Dopaminergic	OPRM1	rs1799971	AA	AA	
Serotoninergic	1A-HTR1A	rs6295	CC	CG	
Serotoninergic	SLC6A4	rs1042173	AA	CC	
GABA	GABRA2	rs279858	TT	TC	
Endocannabinoid	CNR1	rs2023239	TT	TT	
Endocannabinoid	FAAH	rs324420	CC	CC	
Cell signalling	AKT1	rs2494732	TT	TC	
Cell signalling	ANK3	rs1938526	AA	AA	
Cell signalling	ANK4	rs10994336	CC	CC	
Cell signalling	BDNF	rs6265	CC	CC	
Cell signalling	CACNAI	rs1006737	GG	AA	
Cell signalling	CHRNA3	rs16969968	GG	GA	
Cell signalling	CHRNA5	rs16969968	GG	GA	
Wnt signalling	APC	rs2546110	AA	GG	
Wnt signalling	APC	rs3846716	AA	GG	
Wnt signalling	GSK3B	rs334555	CC	CC	
Wnt signalling	GSK3B	rs11925868	AA	CA	
Wnt signalling	CSNK1A1	rs10045427	AA	AC	
Stress response	FKBP5	rs1360780	CC	CC	
Stress response	OXTR	rs53576	GG	AG	
Metilation	MTHFR	rs1801131	CC	TT	
Metilation	MTHFR	rs1801133	CC	AA	
Metilation	MTR	rs1805087	AA	AA	
Neuroinflammation	IL1A	rs17561	GG	CA	
Neuroinflammation	IL1A	rs1800587	CC	GA	
Neuroinflammation	IL1B	rs1143634	CC	GG	
Neuroinflammation	IL1B	rs16944	AA	GG	



Neuroinflammation	ILIRN	rs419598	CC	Π	
Neuroinflammation	IL-6	rs1800795	GG	CG	
Neuroinflammation	IL-10	rs1800896	GG	TC	
Neuroinflammation	TNFA	rs1800629	GG	GG	
Neuroinflammation	CD33	rs3865444	TT	CC	
Neuroinflammation	MS4A	rs744373	TT	AA	
Neuroinflammation	TREM2	rs143332484	CC	CC	
Neuroinflammation	CRP	rs1205	CC	TT	



# **GLOSSARY**

- Fatty acids: molecules consisting of lipids that are formed from a linear hydrogencarbon chain.
- Adenylyl cyclase: an intracellular enzyme that catalyzes the conversion of adenosine triphosphate (ATP) to cyclic adenosine monophosphate (cAMP), playing an essential role in the activation of membrane receptors.
- **DNA:** abbreviation for deoxyribonucleic acid. A molecule present in our cells that contains the genetic information necessary for the development and proper functioning of living organisms.
- Allele: each of the alternative forms of a gene, which may have differences in their sequence.
- Amide: an organic compound that results from replacing a hydrogen atom of ammonia or amines with an acyl.
- Amino acid: base unit that acts as the fundamental structure of proteins.
- Astrocyte: a stellate-like cell belonging to the neuroglia of the central nervous system.
- Cell: basic structural and functional unit of life.
- Synaptic space: space between the end of a neuron and another cell.
- Phosphorylation: phase of the transformation of glucose to glycogen.
- Gene: DNA segment representing the unit of hereditary information.
- **Heterozygous:** when the two alleles of the same gene are different.
- Homozygous: when the two alleles of the same gene are the same.
- Microglia: cells of the central nervous system that function as elements of the immune system, protecting the organism from external and internal aggressions.
- **Neuromodulator:** a type of nerve neurotransmitter involved in modulating the functioning of various brain structures.
- **Neurotransmitter:** a chemical found in the brain and central nervous system that plays a key role in communication between nerve cells or neurons.
- Peptide: molecule containing two or more amino acids.
- **G protein:** family of proteins characterized by the binding of GTP (Guanosine triphosphate) and its subsequent hydrolysis to GDP (Guanosine diphosphate) during its functional cycle.



## **TECHNOLOGY**

**DNA Microarray** technology consists of a solid surface with microscopic reactions (microreactions) or DNA chip, on which molecular probes are attached to detect the presence of target DNA molecules. Probe-target hybridization is usually detected and quantified by measuring the intensity of a specific fluorescence provided by the molecular probe in the samples. This type of technology allows the detection of thousands of specific DNA fragments present in a DNA sample. On the other hand, the specificity in terms of DNA sequence recognition is very high, since single nucleotide exchange (single base resolution) can be detected using short oligonucleotide probes (20-25 nucleotides). As a result, DNA Microarray technology has also evolved to be applied as a DNA sequencing technique to genotype several hundred thousand single nucleotide variants (SNVs) in target genes located throughout the genome (Whole Genome DNA Microarray).

# **QUALITY**

The analytical laboratory has standard and effective procedures to protect against technical and operational problems. However, results can be altered due to problems with sample collection (contamination) and labeling (identification), delay in receiving the sample in the laboratory (integrity), among other problems. This could lead to invalidation of the test results. In such cases, the patient would be asked to repeat the entire testing process.

As with all clinical screening tests, there is a small chance that the laboratory may report inaccurate information. If there is a suspicion of an error in the genotype detected, a verification test may be requested.



Genetics for people



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